

Supplemental Table 1. Genomewide CNV results for unaffected parents of probands with 22q11.2DS

| Unaffected parents with confirmed de novo 22q11.2 deletion status | | | | | | |
|---|------------------|---|--|-------------------------------|----|------|
| | All (n = 118) | Source of de novo 22q11.2 deletion (n = 53) | Not involved in de novo deletion 22q11.2 occurrence (n = 65) | De novo source vs. non-source | | |
| Total number of CNV | 652 | 288 | 364 | | | |
| Stringent * | 283 (n=107) | 128 (n=49) | 155 (n=58) | | | |
| | | | | Statistic | df | p |
| CNV per genome | 5.5 | 5.4 | 5.6 | | | |
| Stringent * | 2.6 | 2.6 | 2.7 | $z = -0.13$ | - | 0.9 |
| Median Size in kb (range) | 194 (2.3 – 2731) | 194 (2.4 – 1987) | 194 (3.0 – 2731) | | | |
| Stringent * | 211 (3.3 – 2731) | 247 (8.2 – 1987) | 198 (3.3 – 2731) | $z = 1.03$ | - | 0.3 |
| >1 Mb CNV (% of total) | 70 (10.7%) | 31 (10.7%) | 39 (10.7%) | | | |
| Stringent | 46 (16.3%) | 21 (16.4%) | 25 (16.1%) | $\Pi^2 = 0.004$ | 1 | 0.95 |
| Losses, % of total | 37.7% | 37.5% | 37.9% | | | |
| Stringent * | 40.3% | 38.3% | 41.9% | $\Pi^2 = 0.39$ | 1 | 0.53 |

* Analysis using stringent data set, i.e., as called by >1 algorithms or arrays. See Methods for further details.